Down Syndrome

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In the world domain, the human species is faced with many challenges. These challenges can be classified under natural disasters or man-induced disasters. If we were to look at epidemics, we can say that the human species dwells in a world where at one time or the other one can to be attacked by an abnormal condition. Abnormal conditions negatively affect some part or the whole of an organism, preventing it from operating at maximum potential. When an organism experiences abnormal conditions it is said to be sick, or diseased. There are many disorders in the world. Some are manageable while others are not. For example, ever since the discovery of HIV/AIDS, no cure has been engineered yet. However, the disease can be contained through medication, although it is not curable once it has attached itself to an organism. The purpose of this paper is to discuss Down Syndrome, a condition that affects intellectual and physical growth in humans, as well as sometimes hindering the development of some facial features.

To understand this concept better, it is best to determine the difference between a disease and a syndrome. In definition, a syndrome is said to be a collection of medical characteristics or symptoms, which are usually ongoing. This means that the physical cause of these symptoms or medical characteristics cannot always be identified. The underlying causes of many syndromes are still unknown, and a measurable anatomical alteration does not always exist (NDSS). In some cases, when the actual cause and full effect of a syndrome is understood, it ends up being classified as a disease. Therefore, from this definition of a syndrome, one can clearly state that a disease is a condition that harms the body's functions, and its underlying cause is known. This

means that a disease is not a medical mystery for its consistent set of symptoms are known; hence has treatment options if any exists.

Therefore, one can say that the main difference between a syndrome and a disease is that while syndromes are still medical mysteries, diseases are more cut and dry and have treatment options. The most interesting relationship between syndrome and diseases is the fact that some diseases can cause syndromes. This is because some diseases create a collection of symptoms which can be considered as syndromes. However, not all disease causes syndromes. Syndromes have many causes including accidents and mental disorders among many other factors. The two terminologies (disease and syndrome) can sometime overlap while investigating some health issues (Difference Between.net).

In definition, the Down Syndrome, which is also known as the Trisomy 21 disorder, is a genetic disorder which occurs when there is the presence of a partial, or a whole extra third copy of Chromosome 21 (MedlinePlus). This disease syndrome is brought about by the fact that every cell found in the human body has a nucleus. All genetic materials are stored in structures called genes. Genes are responsible for carrying genetic codes for all inherited traits from one's parents (March of Dimes, 2009). These traits are grouped within rod-like structures called the chromosomes. It has been scientifically proven that each nucleus in the human body contains 23 pairs of chromosomes, and each half is inherited from a particular parent. However, sometimes there may exist a partial copy or a full extra pair of chromosome 21 during child formation, and this is how the Down Syndrome occurs. This additional genetic material changes the course of that child's development, and causes characteristics which are associated with this syndrome. There are several physical and mental traits that are associated with Down Syndrome (Genetics Home Reference).

The most common physical characteristics of the disease include: a small stature, an upward slant to the eyes, a lower muscle tone than usual, a deep crease running across an individual's center of the palm, and many others. However, these characteristics may not appear all together in an individual for the syndrome tends to show unique traits in every individual. Individuals may also possess these traits at different degrees while others may not even show them at all (CDC, 2014).

The Down Syndrome was a common occurrence in ancient times. It was even documented in the early times in works of science, art and literature. A good example is found in the infamous "*The Adoration of the Christ Child*" painting from the early 16th century (Dobson, 2003). The syndrome is still a common occurrence to date with statistics showing that in every 691 babies in the United States, one is born with the Down Syndrome. This makes Down Syndrome the most common genetic condition in the United States of America. Additionally, statistics show that around 6,000 infants are born with Down Syndrome every year. This number accounts for the approximately 400,000 United States citizens presumed to have the syndrome today (NIH, 2014).

For many centuries, ancient scientists did not know what the syndrome was, and it was mostly characterized as a mental illness. It was in the late nineteenth century when an English physician by the name of John Langdown Down published an article with an accurate description of an individual who possessed characteristics of this syndrome (Ananya Mandal). John Down's scholarly work was published in early 1866, earning him the recognition as the "father" of a syndrome which was considered to be a mental illness. During the discovery of this syndrome, Down was of the perception that people who showed its characteristics possessed similar characteristics to people of the Blumenbach's Mongolian race. As a result, Down used the term

"mongoloid," which continued to be used as the acceptable term until in the early 1970s. Some scientists as well as the Mongolian people felt that the term "mongoloid," which meant Mongolian imbecility, was an unacceptable term for a disease for it was embarrassing to the people of Mongolia. There were also a couple of scientists who felt that the name "mongoloid" possessed some misguided conception for the syndrome had nothing to do with Mongolians. It occurred in every race worldwide regardless of the people's origin (DSE, 2014). As a result, a Mongolian delegate requested the World Health Organization (WHO) to drop the term due to its "misleading connotations." Consequently, the term was dropped by W.H.O in 1965. In 1975, a conference was convened by the National Issues of Health in the United States with the sole aim of standardizing the name of the syndrome. As a result, two terminologies which are frequently used to describe the disease were born: that is "Trisomy 21" and "Downs Syndrome."

In recent times, there have been many more developments into the Down Syndrome due to advancements in medical technology. A good example would be the discovery of the 1950s karyotype technique which made it possible to identify chromosomal shapes and their numbers. As a result, a French physician, Jerome Lejeune, was able to identify the syndrome as a chromosomal condition. Lejeune discovered that instead of an individual having 46 chromosomes in each cell, there were 47 chromosomes in individuals with the condition (Dsaco, 2015). This discovery was later advanced and it was determined that this additional chromosome existed either partially or as a whole copy of chromosome 21. Scientists then went ahead to do more research on chromosome 21 and successfully catalogued the 329 genes found on human chromosomes as a means of understanding the syndrome even better (Pace, 1994). Down Syndrome exists in three different types. They include trisomy 21(also known as nondisjunction) translocation and mosaicism (CDC, 2014).

Trisomy 21 (nondisjunction)

In Trisomy 21, scientists proved that Down Syndrome is usually as a result of a cell division error called "nondisjunction." Consequently, instead of an embryo having the two usual copies of chromosome 21, it inhibits three copies (NIH). This is mostly at, or prior to, conception where a pair of chromosome 21 in either the egg or the sperm fails to separate. Therefore, the extra chromosome ends up being replicated in all cells of the body of a fetus when the embryo is developing. This is the most common type of the Down Syndrome for it accounts for at least 95 percent cases of the syndrome; hence " Trisomy 21."

Translocation

In Translocation, events are totally different at chromosome 21 from the ones in nondisjunction. In this case, instead of an error in cell division in chromosome 21, a part of this chromosome practically breaks off and then attaches to another chromosome. The part typically attaches itself to chromosome 14 after breaking off from chromosome 21 (NDSS, 2015). This means that since there will be an extra part of chromosome 21, even when chromosomes in a cell remain 46, an individual will definitely exhibit characteristics of Down Syndrome (Down syndrome Symptoms). This translocation is responsible for at least 4 percent of all Down Syndrome cases (NDSS).

Mosaicism

In Mosaicism, events are totally different from what happens in translocation, but not necessarily in nondisjunction. This is because; Mosaicism takes place "when nondisjunction occurs in one –but not all- initial cell divisions after fertilization." This means that there is a mixture of two cell types when this occurs. Some of these cells will contain 47 chromosomes

while other cells will contain the usual 46 chromosomes. Those cells with 47 chromosomes are the ones with an extra chromosome 21. This type of Down syndrome accounts for only 1 % of individuals showing the syndrome's characteristics (American Pregnancy Association). As a matter of fact, research has also shown that individuals with Mosaicism show the least of the syndrome as to when compared with individuals with nondisjunction and translocation types (NDSS). However, due to the wide range of capabilities inhibited by individuals with Down Syndrome, broad generalizations are usually not possible.

Down Syndrome has a common cause regardless of the type an individual may inhibit, which is an extra part of chromosome 21, or the whole of it in their nucleus. This partial or full extra chromosome 21 is either present in all of the cells in an individual's body or in some of them (NDSS). This definitely means the presence of the unaccountable genetic material which alters the process of an embryo's development. This extra material causes the characteristics associated with the Down Syndrome (American Pregnancy Association). The cause of nondisjunction is still unknown to scientists. However, research suggests that the frequency of this syndrome increases as a woman ages. However, it appears to be contradicted by the fact that at least 80 percent of children born with this disease are to women younger than 35 years of age. Some scientists have argued that this is because of high birth rates for women between the ages of 18 and 35 years of age.

However, there has been no definitive research that shows any environmental factors or parents' activities during or before pregnancy contribute to this disease (NHS, 2015). Research also suggests that the extra partial or full copy of chromosome 21 responsible for the Down Syndrome can originate from either the mother or the father. Five percent of the Down Syndrome cases have been traced to the father as the source of the partial or the full copy of

chromosome 21. With 15 percent of cases being traced back to the mother as a carrier of Down Syndrome. Older fathers over 40 had twice the rate of Down syndrome births compared with men 24 years old and younger when they had children with women over 35.

Signs and symptoms of Down Syndrome

Down Syndrome has a couple of signs and symptoms. For starters, individuals with this syndrome nearly always have intellectual and physical disabilities. At adulthood, individuals with this syndrome usually have mental abilities which are similar to an 8 or 9 year old with an IQ of about 50. However, the IQ of an individual varies widely, more so depending on whether an individual with the syndrome is educated or not. Individuals with Down Syndrome also tend to have poorer immune functions when compared to people without the syndrome (The Arc, 2011). Additionally, they tend to reach certain developmental milestones at a significantly later stage. Research shows that they also tend to exhibit increased risk in other health matters including leukemia, congenital heart defects, mental illnesses and thyroid disorders among others (NIH, 2014). Below is a table showing a number of physical characteristics exhibited by individuals with Down Syndrome in respect to abnormality percentages.

Characteristics	percentage	Characteristics	percentage
Stunted growth	90%	Slanted eyes	60%
Mental impairment	99%	Short neck	60%
Low muscular tone	80%	Bent fifth finger tip	57%
Flat head	75%	Abnormal teeth	60%
Umbilical hernia	90%	Protruding tongue	47%
Large tongue	75%	Shortened hands	60%
Flattened nose	68%	Obstructive sleep apnea	60%
Increased skin back of neck	80%	Undescended testicles	20%
Separation of first and second toes	68%	Congenital heart defects	40%

When it comes to intellectual matters, individuals with Down Syndrome sometimes prove to be difficult to deal with and also to live within a normal household (Genetics Home Reference). They require constant care like the one accorded a 9 year old, even at old age. This is because most individuals with this syndrome have a moderate IQ of between 35-40, while other individuals have a mild IQ of about 50 -70. Individuals with trisomy 21 are the most affected with severe cases sometimes registering a very low IQ of about 20-35. Individuals with mosaic Down Syndrome always do better than their trisomy 21 and translocation counterparts. This is because they register a higher IQ with about 10-30 points higher in most cases. As people age, individuals with the Down Syndrome tend to perform less when compared to their peers. This is also shown in many classrooms where affected individuals exhibit many learning disabilities and characteristics (Down Syndrome Education International).

There is no definite reference to a single learning disability. This is because there are several classifications of learning disabilities that face mankind. These classifications include different levels at which a Down Syndrome patient is unable to learn in a typical manner. These forms of disability can be caused by one or several unknown factors. As a result, the terminology learning disabilities is used to refer to diverse learning problems in an academic perspective. This is for example; speech skills, language, and other academic challenges. The most common types of academic disabilities in Down syndrome are: dyscalculia (mathematics disability), dyslexia (reading disabilities) and dysgraphia also known as writing disabilities. These types of learning disorders are known, but the factor causing these disorders and the brain's inability to receive and process data remains a mystery (Wilson, 2012). This is attributed to the fact that the underlying causative factor of the Down syndrome remains a mystery. The only known fact about the syndrome in terms of its spreading is that it is genetic.

There are learning disabilities that manifest themselves in two ways. That is, a disorder can make an individual be significantly slow in receiving and processing information more than a normal individual unaffected by the syndrome in question. However, Down Syndrome individuals will achieve the physical development as other children, but on their own timetable. The following chart illustrates milestones achieved within a time frame.

Milestone	Range for Children with Down Syndrome	Typical Range
GROSS MOTOR		
Sits Alone	6 – 30 Months	5 – 9 Months
Crawls	8 – 22 Months	6 – 12 Months
Stands	1 – 3.25 Years	8 – 17 Months
Walks Alone	1 – 4 Years	9 – 18 Months
LANGUAGE		
First Word	1 – 4 Years	1 – 3 Years
Two-Word Phrases	2 – 7.5 Years	15 – 32 Months
SOCIAL/SELF-HELP		
Responsive Smile	1.5 – 5 Months	1 – 3 Months
Finger Feeds	10 – 24 Months	7 – 14 Months
Drinks From Cup Unassisted	12 – 32 Months	9 – 17 Months
Uses Spoon	13 – 39 Months	12 – 20 Months
Bowel Control	2 – 7 Years	16 – 42 Months
Dresses Self Unassisted	3.5 – 8.5 Years	3.25 – 5 Years

(NDSS)

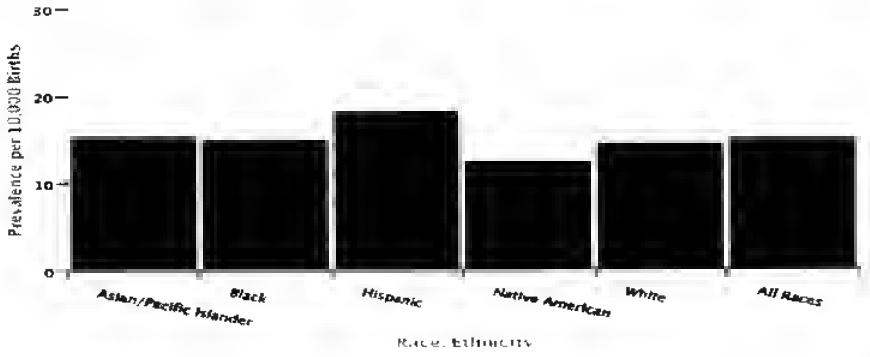
On the other hand, a syndrome can also make an individual able to learn and process specific information much more quickly than a normal individual unaffected by the disease. Most of the times, the disorder is a significant challenge to affected individuals especially the ones taught conventionally, or if entrusted with a task to figure things out individually. However, in the cases where an individual processes information quicker than a normal human being, it leads to giftedness of some kind. As a result, that is why we see some students being exceptionally good in some subjects like mathematics and fail miserably in others subjects like language, and vice versa (Down Syndrome Education International). This also means that such a student will definitely be interested in the subject they understand better than the rest.

Unfortunately, the latter is only a dream for individuals with Down Syndrome for their low IQ always works against them.

Most of these learning disabilities have been associated with other disorders such as the Asperger syndrome and the Down Syndrome. The Asperger disorder is believed to involve alarming delays on skill developments in children, more so when it comes to the most basic ones. This disorder is believed to hinder growth of skills more in three essential categories. The syndrome is believed to involve delays in communication, ability to socialize with others freely and also the ability to use one's imagination (NIH, 2014). The Asperger Syndrome is seen as a very close cousin of the Down Syndrome.

What are the chances of a bearing a child with Down syndrome?

Down syndrome occurs everywhere across the world and it does not affect one race any differently from the other (CDC, 1994). Here is a chart of how Down syndrome in different races.



However, the age of a mother is a totally different thing. This is because the Down Syndrome occurs in older women having children more than in younger women. This is of course when one factors out the high birth rates of women under the age of 35 and factors in the

frequency of Down Syndrome cases recorded in mother of above age 40 and above. As a matter of faet, research has shown that a woman below 35 years of age has just about 1 in 350 chances of bearing a child with Down Syndrome. These chances increase with time, and at the age of 40 years, they become 1 in about 100. By the time a woman is at age 45 years, chances of bearing a child with Down Syndrome increase to as high as 1 chance in 30 (NDSS). According to National Down Syndrome Society, these chances are more so associated with trisomy 21, because the age of a mother has no proven scientific risk to translocation of mosaicism types of Down Syndrome. Below is a table showing the likelihood of bearing a child with Down Syndrome in relation to the age of the mother.

Maternal age	**Incidence of Down Syndrome**	**Maternal age**	**Incidence of Down Syndrome**	**Maternal age**	**Incidence of Down syndrome**
20	1 in 2,000	30	1 in 900	40	1 in 100
21	1 in 1,700	31	1 in 800	41	1 in 80
22	1in 1,500	32	1 in 720	42	1 in 70
23	1 in 1,400	33	1 in 600	43	1 in 50
24	1 in 1,300	34	1 in 450	44	1 in 40
25	1 in 1,200	35	1 in 350	45	1 in 30
26	1 in 1,100	36	1 in 300	46	1 in 25
27	1 in 1,050	37	1 in 250	47	1 in 20
28	1 in 1,000	38	1 in 200	48	1 in 15
29	1 in 950	39	1 in 150	49	1 in 10

Scientists say that the incidence of Down Syndrome are bound to increase in society since many couples are postponing parenting till late in life in chase of career success. A study found that the number of Down Syndrome cases among parents over 40 was 60 per 10,000 births, which is six times more than the rate found among couples under 35 years old. Older fathers over 40 had twice the number of Down Syndrome births then men 24 years old and younger when they had children with women over 35 (New York Presbyterian, 2003).

As a result, topics such as genetic counseling are becoming significantly important in the society so as to educate many people on how to avoid disorders that hide behind the curtain of age. However, research shows that many physicians still do not fully understand the incidences, advancements in diagnosis as well as the right protocols for caring for a child born with Down Syndrome (NDSS).

Due to the fact that Down Syndrome is genetic, there have been numerous concerns whether it runs in families. All three types of the syndrome are genetic conditions meaning that they are all related to genes. However, recent scientific studies in the syndrome seem to show that only one percent of all scenarios of the syndrome have a hereditary component (NDSS). That is only one percent that is passed down to a child from a parent through genes. Scientific studies have also shown that neither mosaicism nor nondisjunction (trisomy 21) is a hereditary factor. However, the same cannot be said for the translocation type of Down Syndrome. This is because when research was carried out when genes were isolated, the extra 47^{th} chromosome showed some degree of hereditary component, which accounts for the 1% cases of the syndrome. These cases seem to be by mere chance, and are in no way connected to the age of the mother. However, one parent is always a carrier in about one third in all translocated chromosome cases (Mayo Clinic).

There is also a significant concern about giving birth to a second child with Down Syndrome after a first case. Research into the matter shows that chances of a woman giving birth to a baby with either translocation of nondisjunction Down syndrome are 1 in 100 (NDSS). This is until the mother is above age 40 where the chances of giving birth to another baby with the same syndrome starts falling to about 1 case in 80 by the time she is 41 years of age. However, when the father is a carrier in a translocation scenario, chances of giving birth to a second child with Down Syndrome are 3 percent, while they are as high as 15 percent if the mother is the carrier (American Pregnancy Association). Research has shown that genetic counseling can assist in determining the source of the translocation type of Down Syndrome.

Diagnosis of Down Syndrome

Down Syndrome can be diagnosed both at birth and prenatally. Prenatally, there are two ways in which a baby can be determined whether they have Down syndrome or not. This can be achieved through screening tests and diagnostic tests. In screening tests, a mother is taken through some screening tests which help in estimating whether a fetus may have Down Syndrome or not. Even with the recent developments in technology and medical science, these screens only provide a probability into the matter. However, diagnostic tests sometimes provide a much better definitive diagnosis of around 99 percent accuracy. Most screening tests involve a sonogram (an ultra sound) and a blood test. The serum screening tests, the blood tests, measure quantities a wide range of substances in a mother's blood. The information obtained from these tests together with the mother's age are used to determine the chances of a child having the syndrome (NIH, 2014).

These serum screening tests are usually done together with a well detailed sonogram to look for any "markers" that may indicate Down Syndrome. These "markers" would be any traits

that physicians may suspect to have any tangible association with Down Syndrome. With the recent developments in technology however, new prenatal screens can now detect chromosomal material circulating in the maternal blood from the fetus. Although these tests do not definitely diagnose the syndrome, they offer a high accuracy rate in the matter. As a matter of fact, pregnant women are strongly advised to take these tests when they are going for clinic checkups. In a developed country like the United States of America, diagnostic and screening tests are offered routinely to all pregnant women regardless of their ages (NIH, 2014).

Other than the screening tests, there are also prenatal diagnostic tests to determine whether an expectant mother carries a child with Down Syndrome or not. Diagnostic procedures involve amniocentesis and chorionic villus sampling (CVS). These procedures have been considered as invasive by some people because they carry at least a 1 percent chance of a miscarriage. However, their results are 100 percent accurate on whether the baby has Down Syndrome or not. The chorionic villus sampling procedure is carried out between 9 and 11 weeks of pregnancy. The amniocentesis procedure is usually carried out after 15 weeks of gestation, which is in the second trimester (Kids Health).

A baby can also be determined whether it carries Down Syndrome at birth. At this time it is usually much easier for a physician to tell whether a baby has the syndrome because of visible physical features. A baby with Down Syndrome exhibits features such as upward slanted eyes, a single transverse palmar crease, slightly flattened facial profile as well as low muscle tone. However, since these features may also be present in babies without Down Syndrome, a procedure called karyotype, which determines the number of chromosomes in the baby's cells is performed. A physician simply uses a blood sample for examining the baby's cells, and from the test he can tell whether it has Down Syndrome or not. In a karyotype test, doctors use

chromosome sizes, numbers and shapes from photographs to test for the presence of Down Syndrome. Nowadays there is even a much faster test called "FISH" that uses similar principles in confirming the diagnosis (Morgan Stanley Children's Hospital).

Management of Down Syndrome

There are several practices that help in management of Down Syndrome. For instance, research has shown that practices such as screening for common problems in children with the syndrome and early childhood intervention helps in managing the effects of the condition. A good family environment, work and educational-related training and medical treatment where deemed necessary by a qualified physician in the field can also help in improving the quality of life of an individual. Screening for problems invisible to the human eye such as congenital heart problems, can be corrected from as early as three months of age and helps in improving the management of a child with the condition (American Pregnancy Association).

There is also cognitive development which helps in improving the development of an individual with the syndrome. Cognitive development can be achieved through practices such as speech therapy, teaching individuals sign language, or even buying hearing aids for a kid whose hearing may be impaired for language learning (NDSS). There are also other management practices such as Tonsillectomy, a procedure to help kids with throat infections from the syndrome as well as sleep apnea. Physical therapy can also be used to improve an individual's motor skills especially when it comes to adults with Down Syndrome. Other than these medical means of managing Down Syndrome, there are also other practices which are poorly supported by evidence. However, scientists remain optimistic that once the underlying cause of the extra chromosome 21 causing the syndrome is determined, a proper cure would be engineered (Mayo Clinic, 2014).

Impact of Down Syndrome on society

History shows that in the ancient times, individuals with Down Syndrome were significantly despised in the society. Parents who fathered children with this condition would most often abandon them while others would even kill their own flesh and blood (Ananya Mandal). However, thanks to civilization the world is now much better informed. Individuals with Down Syndrome have become widely accepted in the society, and they have even been integrated in community organizations such as schools, work forces, healthcare systems as well as in social and recreational activities. This is even when they show varying degrees of cognitive delays which can range from very mild to significantly severe (NHS, 2015).

Additionally, advancements in medical technology more so in the 21st century have ensured that individuals with this syndrome can now live up to ages of about 50 and 60 years of age. As a matter of fact, research shows that around 1910, individuals with the syndrome were only expected to live for about 8 to 10 years. When antibiotics were discovered several decades later, their life span improved to around 18 or 20 years. Nowadays, things are totally different thanks to clinical treatment advancements, more so in the field of corrective heart surgeries. Research shows that 80 % individuals who are well taken care of in the society get to reach age 60 and sometimes even longer (NDSS). These are impressive statistics bearing in mind where the world was a century ago, and with more education and acceptance of the condition in the society, the world can conquer the syndrome as long as scientists continue researching into the matter.

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